Anita Rauch

List of Publications by Year
in descending order

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255	17,020	17776 65	22488 117
papers	citations	h-index	g-index

270	270	270	25224
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	6.3	940
2	Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336.	9.4	670
3	CNTNAP2 and NRXN1 Are Mutated in Autosomal-Recessive Pitt-Hopkins-like Mental Retardation and Determine the Level of a Common Synaptic Protein in Drosophila. American Journal of Human Genetics, 2009, 85, 655-666.	2.6	573
4	Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. Cell, 2008, 135, 37-48.	13.5	567
5	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	9.4	431
6	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
7	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	6.0	370
8	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 2063-2074.	0.7	343
9	Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. American Journal of Human Genetics, 2007, 80, 550-560.	2.6	316
10	Contribution of Global Rare Copy-Number Variants to the Risk of Sporadic Congenital Heart Disease. American Journal of Human Genetics, 2012, 91, 489-501.	2.6	272
11	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001.	2.6	261
12	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	9.4	236
13	Genotype-phenotype correlations in Noonan syndrome. Journal of Pediatrics, 2004, 144, 368-374.	0.9	227
14	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. American Journal of Human Genetics, 2012, 90, 565-572.	2.6	225
15	The core FOXG1 syndrome phenotype consists of postnatal microcephaly, severe mental retardation, absent language, dyskinesia, and corpus callosum hypogenesis. Journal of Medical Genetics, 2011, 48, 396-406.	1.5	220
16	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	9.4	201
17	Human TBX1 Missense Mutations Cause Gain of Function Resulting in the Same Phenotype as 22q11.2 Deletions. American Journal of Human Genetics, 2007, 80, 510-517.	2.6	195
18	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	6.0	195

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19	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. Journal of Medical Genetics, 2009, 46, 598-606.	1.5	194
20	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720.	1.5	191
21	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	1.4	190
22	Identification of a novel loss-of-function calcium channel gene mutation in short QT syndrome (SQTS6). European Heart Journal, 2011, 32, 1077-1088.	1.0	178
23	Induction, binding specificity and function of human ICOS. European Journal of Immunology, 2000, 30, 3707-3717.	1.6	166
24	Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. Human Mutation, 2010, 31, 722-733.	1.1	163
25	Mutations at the SALL4 locus on chromosome 20 result in a range of clinically overlapping phenotypes, including Okihiro syndrome, Holt-Oram syndrome, acro-renal-ocular syndrome, and patients previously reported to represent thalidomide embryopathy. Journal of Medical Genetics, 2003, 40, 473-478.	1.5	159
26	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	2.6	151
27	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	2.6	151
28	Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. European Journal of Human Genetics, 2004, 12, 879-890.	1.4	149
29	Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963.	1.4	147
30	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	9.4	146
31	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	1.4	144
32	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	1.5	135
33	Altered TGFÎ² signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 deficiency. European Journal of Human Genetics, 2010, 18, 895-901.	1.4	132
34	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
35	First known microdeletion within the Wolf-Hirschhorn syndrome critical region refines genotype-phenotype correlation. American Journal of Medical Genetics Part A, 2001, 99, 338-342.	2.4	128
36	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331.	1.5	126

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37	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
38	?Mowat-Wilson? syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomalies-mental retardation syndrome caused by mutations in the zinc finger homeo box 1B gene. American Journal of Medical Genetics Part A, 2002, 108, 177-181.	2.4	122
39	Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111.	0.7	121
40	Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. Journal of Medical Genetics, 2005, 42, 871-876.	1.5	118
41	Severely Incapacitating Mutations in Patients with Extreme Short Stature Identify RNA-Processing Endoribonuclease RMRP as an Essential Cell Growth Regulator. American Journal of Human Genetics, 2005, 77, 795-806.	2.6	117
42	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. Journal of Medical Genetics, 2007, 44, 651-656.	1.5	114
43	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106.	2.1	109
44	Molecular karyotyping using an SNP array for genomewide genotyping. Journal of Medical Genetics, 2004, 41, 916-922.	1.5	106
45	Phenotype-specific effect of chromosome 1q21.1 rearrangements and GJA5 duplications in 2436 congenital heart disease patients and 6760 controls. Human Molecular Genetics, 2012, 21, 1513-1520.	1.4	101
46	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	1.1	101
47	A novel microdeletion syndrome involving 5q14.3-q15: clinical and molecular cytogenetic characterization of three patients. European Journal of Human Genetics, 2009, 17, 1592-1599.	1.4	96
48	A Novel 22q11.2 Microdeletion in DiGeorge Syndrome. American Journal of Human Genetics, 1999, 64, 659-667.	2.6	95
49	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491.	3.9	89
50	Gene discovery for Mendelian conditions via social networking: de novo variants in KDM1A cause developmental delay and distinctive facial features. Genetics in Medicine, 2016, 18, 788-795.	1.1	88
51	Further delineation of Pitt-Hopkins syndrome: phenotypic and genotypic description of 16 novel patients. Journal of Medical Genetics, 2008, 45, 738-744.	1.5	86
52	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.5	84
53	Microcephalin and pericentrin regulate mitotic entry via centrosome-associated Chk1. Journal of Cell Biology, 2009, 185, 1149-1157.	2.3	83
54	Disturbed Wnt Signalling due to a Mutation in CCDC88C Causes an Autosomal Recessive Non-Syndromic Hydrocephalus with Medial Diverticulum. Molecular Syndromology, 2010, 1, 99-112.	0.3	82

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55	Genome-wide association study identifies loci on 12q24 and 13q32 associated with Tetralogy of Fallot. Human Molecular Genetics, 2013, 22, 1473-1481.	1.4	82
56	Deletion mapping on chromosome 10p and definition of a critical region for the second DiGeorge syndrome locus (DGS2). European Journal of Human Genetics, 1998, 6, 213-225.	1.4	81
57	A study of ten small supernumerary (marker) chromosomes identified by fluorescence in situ hybridization (FISH). Clinical Genetics, 1992, 42, 84-90.	1.0	81
58	Type and Level of RMRP Functional Impairment Predicts Phenotype in the Cartilage Hair Hypoplasiaâ€“Anauxetic Dysplasia Spectrum. American Journal of Human Genetics, 2007, 81, 519-529.	2.6	78
59	Crisponi Syndrome Is Caused by Mutations in the CRLF1 Gene and Is Allelic to Cold-Induced Sweating Syndrome Type 1. American Journal of Human Genetics, 2007, 80, 971-981.	2.6	76
60	Allelic Heterogeneity in the COH1 Gene Explains Clinical Variabilityin Cohen Syndrome. American Journal of Human Genetics, 2004, 75, 138-145.	2.6	72
61	Klinefelter syndrome and mediastinal germ cell tumors. American Journal of Medical Genetics, Part A, 2006, 140A, 471-481.	0.7	72
62	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636.	1.5	72
63	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	1.5	72
64	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. European Journal of Human Genetics, 2015, 23, 602-609.	1.4	72
65	Dosage-Dependent Severity of the Phenotype in Patients with Mental Retardation Due to a Recurrent Copy-Number Gain at Xq28 Mediated by an Unusual Recombination. American Journal of Human Genetics, 2009, 85, 809-822.	2.6	70
66	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	1.6	70
67	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	3.7	70
68	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	1.5	67
69	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	1.1	67
70	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	1.5	66
71	Incidence and significance of 22q11.2 hemizygosity in patients with interrupted aortic arch. , 1998, 78, 322-331.		62
72	Characterisation of deletions of the ZFHX1B region and genotype-phenotype analysis in Mowat-Wilson syndrome. Journal of Medical Genetics, 2003, 40, 601-605.	1.5	61

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73	Systematic survey of variants in TBX1 in non-syndromic tetralogy of Fallot identifies a novel 57 base pair deletion that reduces transcriptional activity but finds no evidence for association with common variants. Heart, 2010, 96, 1651-1655.	1.2	61
74	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	1.5	60
75	The face of Noonan syndrome: Does phenotype predict genotype. American Journal of Medical Genetics, Part A, 2010, 152A, 1960-1966.	0.7	59
76	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	2.6	59
77	Hirschsprung disease, mental retardation, characteristic facial features, and mutation in the geneZFHX1B (SIP1): Confirmation of the Mowat-Wilson syndrome. American Journal of Medical Genetics Part A, 2003, 116A, 385-388.	2.4	58
78	Laterality of the aortic arch and anomalies of the subclavian artery?reliable indicators for 22q11.2 deletion syndromes?. European Journal of Pediatrics, 2004, 163, 642-5.	1.3	58
79	In-Frame Deletion and Missense Mutations of the C-Terminal Helicase Domain of <i>SMARCA2</i> in Three Patients with Nicolaides-Baraitser Syndrome. Molecular Syndromology, 2011, 2, 237-244.	0.3	58
80	A new quantitative PCR multiplex assay for rapid analysis of chromosome 17p11.2-12 duplications and deletions leading to HMSN/HNPP. European Journal of Human Genetics, 2003, 11, 170-178.	1.4	57
81	Dosage changes of MED13L further delineate its role in congenital heart defects and intellectual disability. European Journal of Human Genetics, 2013, 21, 1100-1104.	1.4	57
82	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058.	1.1	57
83	Highly variable cutis laxa resulting from a dominant splicing mutation of the elastin gene. American Journal of Medical Genetics, Part A, 2008, 146A, 977-983.	0.7	56
84	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
85	A Dual Phenotype of Periventricular Nodular Heterotopia and Frontometaphyseal Dysplasia in One Patient Caused by a Single FLNA Mutation Leading to Two Functionally Different Aberrant Transcripts. American Journal of Human Genetics, 2004, 74, 731-737.	2.6	55
86	Independent NF1 and PTPN11 mutations in a family with neurofibromatosisâ€Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1263-1267.	0.7	55
87	Mutations in <scp>CDK</scp> 5 <scp>RAP</scp> 2 cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480.	0.6	55
88	Clinical and molecular cytogenetic observations in three cases of â€œtrisomy 12p syndromeâ€• , 1996, 63, 243-249.		54
89	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
90	Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332.	1.1	53

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91	The role of recessive inheritance in early-onset epileptic encephalopathies: a combined whole-exome sequencing and copy number study. European Journal of Human Genetics, 2019, 27, 408-421.	1.4	52
92	The MEF2C-Related and 5q14.3q15 Microdeletion Syndrome. Molecular Syndromology, 2011, 2, 164-170.	0.3	51
93	Lethal cutis laxa with contractural arachnodactyly, overgrowth and soft tissue bleeding due to a novel homozygous fibulinâ€4 gene mutation. Clinical Genetics, 2009, 76, 276-281.	1.0	50
94	A new face of Borjesonâ€“Forssmanâ€“Lehmann syndrome? De novo mutations inPHF6in seven females with a distinct phenotype. Journal of Medical Genetics, 2013, 50, 838-847.	1.5	50
95	Mutations in CRADD Result in Reduced Caspase-2-Mediated Neuronal Apoptosis and Cause Megalencephaly with a Rare Lissencephaly Variant. American Journal of Human Genetics, 2016, 99, 1117-1129.	2.6	50
96	Partial deletion of the critical 1.5 Mb interval in Williams-Beuren syndrome. Journal of Medical Genetics, 2003, 40, 99e-99.	1.5	49
97	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	2.6	48
98	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
99	IntragenicKANSL1mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotypeâ€“phenotype correlations in a large cohort of patients. Journal of Medical Genetics, 2015, 52, 804-814.	1.5	47
100	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	1.4	47
101	Mosaicism for the Charcot-Marie-Tooth disease type 1A duplication suggests somatic reversion. Human Genetics, 1996, 98, 22-28.	1.8	46
102	A variable combination of features of Noonan syndrome and neurofibromatosis type I are caused by mutations in theNF1 gene. American Journal of Medical Genetics, Part A, 2006, 140A, 2749-2756.	0.7	46
103	The molecular basis of the cartilage-hair hypoplasiaâ€“anauxetic dysplasia spectrum. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 131-142.	2.2	46
104	Monosomy 22q11 in patients with pulmonary atresia, ventricular septal defect, and major aortopulmonary collateral arteries. Heart, 1998, 79, 180-185.	1.2	45
105	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	2.6	44
106	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 152 Td (a Report of eight cases including a living child and further evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1268-1281.	0.7	43
107	Microarrays in prenatal diagnosis. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 42, 53-63.	1.4	43
108	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	4.1	43

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109	Mesomelia-Synostoses Syndrome Results from Deletion of SULF1 and SLCO5A1 Genes at 8q13. American Journal of Human Genetics, 2010, 87, 95-100.	2.6	42
110	Highâ€resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. Prenatal Diagnosis, 2014, 34, 525-533.	1.1	42
111	Further corroboration of distinct functional features in SCN2A variants causing intellectual disability or epileptic phenotypes. Molecular Medicine, 2019, 25, 6.	1.9	42
112	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
113	Ichthyosis follicularis, alopecia, and photophobia (IFAP) syndrome: Clinical and neuropathological observations in a 33-year-old man. , 1998, 78, 371-377.		40
114	The shortest of the short: Pericentrin mutations and beyond. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 125-130.	2.2	40
115	Plasma metabolomics reveals a diagnostic metabolic fingerprint for mitochondrial aconitase (ACO2) deficiency. PLoS ONE, 2017, 12, e0176363.	1.1	40
116	Neocentric small supernumerary marker chromosomes (sSMC) â€“ three more cases and review of the literature. Cytogenetic and Genome Research, 2007, 118, 31-37.	0.6	37
117	Analysis of an interstitial deletion in a patient with Kallmann syndrome, Xâ€linked ichthyosis and mental retardation. Clinical Genetics, 1998, 54, 45-51.	1.0	37
118	The smallest teeth in the world are caused by mutations in the PCNT gene. American Journal of Medical Genetics, Part A, 2011, 155, 1398-1403.	0.7	37
119	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37
120	Mowat-Wilson syndrome and mutation in the zinc finger homeo box 1B gene: a well defined clinical entity. Journal of Medical Genetics, 2004, 41, 16e-16.	1.5	36
121	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	1.1	36
122	Autosomal-Dominant Hypertension With Type E Brachydactyly Is Caused by Rearrangement on the Short Arm of Chromosome 12. Hypertension, 2004, 43, 471-476.	1.3	35
123	Pulmonary hypoplasiaâ€“diaphragmatic herniaâ€“anophthalmiaâ€“cardiac defect (PDAC) syndrome due to STRA6 mutationsâ€”What are the minimal criteria?. American Journal of Medical Genetics, Part A, 2009, 149A, 2457-2463.	0.7	35
124	Goltzâ€“Gorlin (focal dermal hypoplasia) and the microphthalmia with linear skin defects (MLS) syndrome: no evidence of genetic overlap. European Journal of Human Genetics, 2009, 17, 1207-1215.	1.4	35
125	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	1.5	35
126	A recurrent germline mutation in the PIGA gene causes Simpsonâ€Golabiâ€Behmel syndrome type 2. American Journal of Medical Genetics, Part A, 2016, 170, 392-402.	0.7	34

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127	Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. European Journal of Medical Genetics, 2017, 60, 451-464.	0.7	34
128	De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. Genetics in Medicine, 2019, 21, 1008-1014.	1.1	34
129	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
130	N⁸â€acetylspermidine as a potential plasma biomarker for Snyderâ€Robinson syndrome identified by clinical metabolomics. Journal of Inherited Metabolic Disease, 2016, 39, 131-137.	1.7	33
131	A novel 5q35.3 subtelomeric deletion syndrome. , 2003, 121A, 1-8.		32
132	AtypicalZFHX1B mutation associated with a mild Mowatâ€“Wilson syndrome phenotype. American Journal of Medical Genetics, Part A, 2006, 140A, 869-872.	0.7	32
133	Unmasking of a Recessive SCARF2 Mutation by a 22q11.12 de novo Deletion in a Patient with Van den Ende-Gupta Syndrome. Molecular Syndromology, 2010, 1, 239-245.	0.3	32
134	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	1.1	32
135	"Mowat-Wilson" syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomalies-mental retardation syndrome caused by mutations in the zinc finger homeo box 1B gene. American Journal of Medical Genetics Part A, 2002, 108, 177-81.	2.4	32
136	A missense mutation in theZFHX1B gene associated with an atypical Mowatâ€“Wilson syndrome phenotype. American Journal of Medical Genetics, Part A, 2006, 140A, 1223-1227.	0.7	31
137	Novel missense, insertion and deletion mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with congenital insensitivity to pain with anhidrosis. Neuromuscular Disorders, 2008, 18, 159-166.	0.3	31
138	Somatic mosaicism in a mother of two children with Pittâ€“Hopkins syndrome. Clinical Genetics, 2013, 83, 73-77.	1.0	31
139	Association Between C677T Polymorphism of Methylene Tetrahydrofolate Reductase and Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2013, 6, 347-353.	5.1	31
140	Facial phenotype allows diagnosis of Mowat-Wilson syndrome in the absence of hirschsprung disease. American Journal of Medical Genetics Part A, 2004, 124A, 102-104.	2.4	30
141	Severe skeletal dysplasia caused by undiagnosed hypothyroidism. European Journal of Medical Genetics, 2007, 50, 209-215.	0.7	30
142	Infantile Epileptic Encephalopathy, Transient Choreoathetotic Movements, and Hypersomnia due to a De Novo Missense Mutation in the SCN2A Gene. Neuropediatrics, 2014, 45, 261-264.	0.3	30
143	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	2.6	29
144	Balanced translocation in a patient with craniosynostosis disrupts the SOX6 gene and an evolutionarily conserved non-transcribed region. Journal of Medical Genetics, 2006, 43, 534-540.	1.5	28

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145	Two novel mutations in the insulin binding subunit of the insulin receptor gene without insulin binding impairment in a patient with Rabsonâ€“Mendenhall syndrome. Molecular Genetics and Metabolism, 2008, 94, 356-362.	0.5	28
146	NovelKIF7mutations extend the phenotypic spectrum of acrocallosal syndrome. Journal of Medical Genetics, 2012, 49, 713-720.	1.5	28
147	LETM1 haploinsufficiency causes mitochondrial defects in Wolf-Hirschhorn syndrome patient cells: implications for dissecting underlying pathomechanisms in this condition. DMM Disease Models and Mechanisms, 2014, 7, 535-45.	1.2	26
148	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17.	1.4	26
149	Inversion Region for Hypertension and Brachydactyly on Chromosome 12p Features Multiple Splicing and Noncoding RNA. Hypertension, 2008, 51, 426-431.	1.3	25
150	Macrocerebellum: Significance and Pathogenic Considerations. Cerebellum, 2012, 11, 1026-1036.	1.4	25
151	A newly recognized 13q12.3 microdeletion syndrome characterized by intellectual disability, microcephaly, and eczema/atopic dermatitis encompassing the HMGB1 and KATNAL1 genes. American Journal of Medical Genetics, Part A, 2014, 164, 1277-1283.	0.7	25
152	Hydrops, fetal pleural effusions and chylothorax in three patients with CBL mutations. American Journal of Medical Genetics, Part A, 2015, 167, 394-399.	0.7	24
153	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	1.1	24
154	Monosomy 1p36 ??? a recently delineated, clinically recognizable syndrome. Clinical Dysmorphology, 2002, 11, 43-48.	0.1	23
155	Mother and daughter with a terminal Xp deletion: Implication of chromosomal mosaicism and X-inactivation in the high clinical variability of the microphthalmia with linear skin defects (MLS) syndrome. European Journal of Medical Genetics, 2007, 50, 421-431.	0.7	23
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